



FIG4 gene

FIG4 phosphoinositide 5-phosphatase

Normal Function

The *FIG4* gene provides instructions for making a protein called Fig4, whose function is not well understood. Researchers believe that the Fig4 protein plays a role in regulating a compound called phosphatidylinositol-3,5-bisphosphate, or PtdIns(3,5)P₂. This compound is involved in the movement of small sac-like structures called vesicles that transport certain substances within cells (vesicle trafficking). The Fig4 protein also seems to help in the survival of nerve cells (neurons).

Health Conditions Related to Genetic Changes

amyotrophic lateral sclerosis

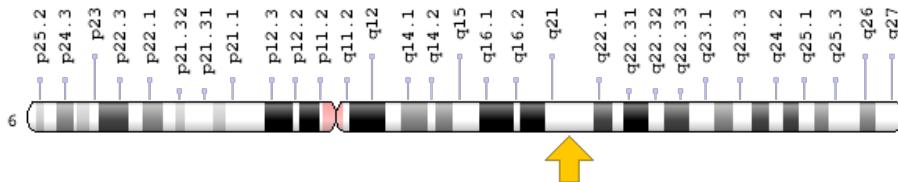
Charcot-Marie-Tooth disease

At least five mutations in the *FIG4* gene have been identified in individuals with Charcot-Marie-Tooth disease type 4J. One of these mutations replaces the protein building block (amino acid) isoleucine with the amino acid threonine at position 41 (written as Ile41Thr or I41T). This mutation has been identified in one copy of the *FIG4* gene in everyone with Charcot-Marie-Tooth disease type 4J. In addition, each individual has another mutation in the other copy of their *FIG4* gene. These mutations change single amino acids in the protein sequence or result in an abnormally short, non-functional protein. Researchers are working to determine how mutations in the *FIG4* gene lead to the specific signs and symptoms of Charcot-Marie-Tooth disease type 4J.

Chromosomal Location

Cytogenetic Location: 6q21, which is the long (q) arm of chromosome 6 at position 21

Molecular Location: base pairs 109,691,216 to 109,825,431 on chromosome 6 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- ALS11
- CMT4J
- dJ249I4.1
- FIG4 homolog (*S. cerevisiae*)
- FIG4 homolog, SAC1 lipid phosphatase domain containing (*S. cerevisiae*)
- hSac3
- KIAA0274
- RP1-249I4.1
- Sac domain-containing inositol phosphatase 3
- SAC3
- SAC3_HUMAN

Additional Information & Resources

GeneReviews

- Charcot-Marie-Tooth Neuropathy Type 4
<https://www.ncbi.nlm.nih.gov/books/NBK1468>

Scientific Articles on PubMed

- PubMed
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28KIAA0274%5BALL%5D%29+OR+%28FIG4%5BALL%5D%29+AND+english%5Bla%5D+AND+%22last+3600+days%22%5Bdp%5D>

OMIM

- FIG4, S. CEREVISIAE, HOMOLOG OF
<http://omim.org/entry/609390>

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
<http://atlasgeneticsoncology.org/Genes/GC FIG4.html>
- ClinVar
<https://www.ncbi.nlm.nih.gov/clinvar?term=FIG4%5Bgene%5D>
- HGNC Gene Family: Phosphoinositide phosphatases
<http://www.genenames.org/cgi-bin/genefamilies/set/1079>
- HGNC Gene Symbol Report
http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=16873
- Inherited Peripheral Neuropathies Mutation Database
<http://www.molgen.ua.ac.be/CMTMutations/Mutations/Mutations.cfm?Context=42>
- NCBI Gene
<https://www.ncbi.nlm.nih.gov/gene/9896>
- UniProt
<http://www.uniprot.org/uniprot/Q92562>

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<https://ghr.nlm.nih.gov/gene/FIG4>

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